RhD Gene (RHD) Copy Number

ARUP test code 005368

RhD Gene (RHD) Copy Number Specimen

Ammniotic fluid

RhD Gene (RHD) Copy Number

1 copy

Indication for testing: Determine fetal RhD zygosity to assess risk for alloimmune hemolytic disease of the fetus and newborn (HDFN).

Heterozygous: One copy of the RhD allele was detected in this prenatal sample, predictive of an RhD-positive phenotype in this fetus.

This result has been reviewed and approved by [Redacted]

BACKGROUND INFORMATION: RhD Gene (RHD) Copy Number

CHARACTERISTICS: Fetal or neonatal erythroblastosis and hydrops. INCIDENCE OF RHD NEGATIVE GENOTYPE: 15 percent Caucasians, 5 percent African Americans, less than 1 percent Asians.

INHERITANCE: Autosomal recessive

CAUSE: Maternal-fetal Rh D antigen incompatibility

CLINICAL SENSITIVITY: Greater than 98 percent.

METHODS: Determine the presence of the RHD exons 5, 7, and a 37 base pair insertion in the intron 3/exon 4 boundary by PCR and fluorescence monitoring. Allelic height ratios are used to determine the number of copies of RHD as compared to RHCE.

ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.

LIMITATIONS: A fetal sample is required for determination of fetal RhD genotype. Bloody amniotic fluid specimens may give false-negative results because of maternal cell contamination; specificity may be compromised by mutations in primer sites or those outside the RHD exons examined; fetuses predicted to be unaffected should continue to be monitored by noninvasive means. Diagnostic errors can occur due to rare sequence variations.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.
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